2026 Midyear Impact Report

Since our founding in 2018, the Kabuki Syndrome Foundation has driven patient-centered research through fundraising and collaboration with partners across the globe. We work every day to improve the lives of those with Kabuki syndrome through treatments, community, and improved clinical care.

$2.8M
awarded in KSF grants across 24 institutions

1,610
people with Kabuki syndrome connected

150
families funded to see an expert and participate in research

Improving Access to Care with the ICD-10-CM Code: Q89.81

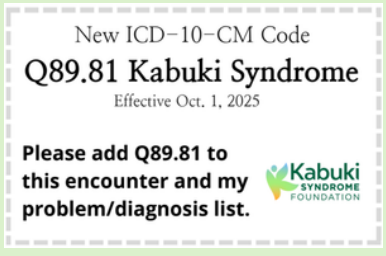
Sometimes improving care isn't about developing a new treatment. It's about removing barriers that stand in the way of families getting the care and support they need today.

Until last October, Kabuki syndrome didn't have a dedicated **ICD-10-CM code**. ICD-10-CM is a system of codes used to label and track information about all conditions and symptoms at medical appointments in the United States. It enables streamlined care and billing nationally, while supporting **data collection that can benefit everyone with Kabuki syndrome.**

Without a dedicated code, families often faced unnecessary challenges obtaining insurance approval for therapies and services. After more than **two years of advocacy**, KSF helped secure a dedicated ICD-10-CM code for Kabuki syndrome: **Q89.81**. And it's already making a difference for families!

Virginia B. wanted occupational therapy for her son Sam, who was facing new challenges due to Kabuki syndrome. Additional private therapies may have previously been denied or delayed, due to the need to use multiple ICD-10-CM codes. But Sam's pediatrician was able to request the additional treatments using the new code, and they were approved.
Now Sam is getting the best care possible. Read more about our code.

"I think the thing about Sam is that he doesn't fit the mold for a kid with 'special needs.' He can walk, talk, learn, and do lots of things, so having the precise diagnostic code is the best way to describe him."
Virginia B., mom to Sam, 9 years old



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Preparing for Clinical Trials with KSOC

With **multiple potential treatment programs** now in development, Kabuki syndrome research is entering an exciting new chapter: clinical trial readiness becomes a top priority.

Being ready means understanding genetic changes and symptoms, having the tools to measure treatment effects, and knowing the families who may be interested in participating.

Meet Emma

Emma, now 12 years old, has Kabuki syndrome and an extreme fear of needles. But she didn't let fear stop her from participating in **KSOC, the Kabuki Syndrome Outcome measures and biomarkers Consortium**. She was so motivated to participate and help us develop tools for measuring the effects of potential treatments in clinical trials that she took a needle phobia class in advance of the blood draw. **Now that's motivation!**

Emma and her family, who live in the Seattle area, were able to participate in KSOC in three short visits at Seattle Children's Hospital. Emma had a blood draw (for which she was prepared!), played cognitive games like puzzles, and had a few other simple tests done. And just like that, **Emma was able to help researchers develop treatments for people with Kabuki syndrome all around the world.**

> *"KSOC was really fun because we got to play games and show the researchers what we know so they can learn more about the Kabuki syndrome genes.* ***It's important to do research so people can learn more about Kabuki syndrome.***"
> Emma, age 12

If you or your loved one has Kabuki syndrome and wants to be notified of new studies like KSOC and future clinical trials, join our global census and contact registry at **www.kabukicount.com**

Every family, researcher, and donor moves us closer to answers for Kabuki syndrome.
Your support makes this report possible. **Thank you.**

Fund More Research & Family Resources:
www.kabukisyndromefoundation.org/donate

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